DOCUMENT RESUME

ED 410 492 CG 027 966

AUTHOR Stebbins, Molly S.; McIntosh, David E.
TITLE The Educational and Psychological Interventions for Children and Adolescents with Marfan Syndrome.
PUB DATE 1997-04-00
NOTE 12p.; Paper presented at the Annual Meeting of the National Association of School Psychologists (Anaheim, CA, April 2-5, 1997).
PUB TYPE Information Analyses (070) -- Speeches/Meeting Papers (150)
EDRS PRICE MF01/PC01 Plus Postage.
DESCRIPTORS Adolescents; Children; Chronic Illness; Elementary Secondary Education; *High Risk Students; *Intervention; Learning Disabilities; *Physical Disabilities; Psychoeducational Methods; *Special Health Problems; Student Needs
IDENTIFIERS *Marfan Syndrome

ABSTRACT

Marfan's syndrome is an autosomal dominant chromosomal disorder of connective tissue which may cause major abnormalities in the musculoskeletal, ocular (pertaining to the eye), and cardiovascular systems of the body. A description of this disorder is presented in this paper. It affects approximately .03 to .05% of the population; approximately 40,000 persons in the United States are currently diagnosed with Marfan's syndrome. Although little research has been conducted exploring the educational and psychological features of the co-presence of other disorders, such as learning disabilities or attention deficit hyperactivity disorder, one study identified 13% of Marfan syndrome sufferers as having a learning disability and 17% as having attention deficit disorders. Although these results may suggest a higher incidence of learning disabilities and attention deficit disorders among individuals with Marfan Syndrome compared to the general population, the author recommended caution in generalizing the results of the entire Marfan population given the small size of their sample and the uncertainty of the incidence of learning disabilities among the general population. Developmental impacts of the syndrome are also poorly understood, although some tentative conclusions can be advanced. Some of the psychoeducational implications are explored here, including school behavior and what steps school psychologists can take to help these children. (RJM)

****
* Reproductions supplied by EDRS are the best that can be made *
* from the original document. *
****

ED 410 492

The Educational and Psychological Interventions for Children and Adolescents with Marfan Syndrome

Molly S. Stebbins and David E. McIntosh
University of Missouri-Columbia

"PERMISSION TO REPRODUCE THIS MATERIAL HAS BEEN GRANTED BY

M. Stebbins

TO THE EDUCATIONAL RESOURCES INFORMATION CENTER (ERIC)."

U.S. DEPARTMENT OF EDUCATION
Office of Educational Research and Improvement
EDUCATIONAL RESOURCES INFORMATION CENTER (ERIC)

☐ This document has been reproduced as received from the person or organization originating it.

☐ Minor changes have been made to improve reproduction quality.

• Points of view or opinions stated in this document do not necessarily represent official OERI position or policy.

Presented at the 1997 Annual Convention of the National Association of School Psychologists, Anaheim, CA, April 2 - 5.

Please address all correspondence to: Dr. David McIntosh
16 Hill Hall
Dept. of Educational and Counseling Psychology
University of Missouri-Columbia
Columbia, MO 65211

The poster was developed from the attached book chapter:

McIntosh, D. E., & Stebbins, M. S. (in press). Marfan Syndrome. In L. Phelps (Ed.), A practitioner's handbook of health-related disorders in children. Washington, DC: American Psychological Association.

CG027966

BEST COPY AVAILABLE

Marfan Syndrome

David E. McIntosh and Molly S. Stebbins

University of Missouri-Columbia

Overview

Antoine Bernard-Jean Marfan at a meeting of the Medical Society of Paris in 1896 first described the characteristics of the syndrome that now bears his name (Tolliss, 1995). An autosomal dominant chromosomal disorder of connective tissue, caused by mutations of the fibrillin-1 gene which is located on chromosome 15 (Aoyama, Francke, Gasner, & Furthmayer, 1995). This mutation results in a defective protein, fibrillin. Fibrillin is a constituent of microfibrils; the later can (in certain tissues) combine with elastin (i.e., protein) to form elastic fibers (Dietz & Pyeritz, 1995). The population prevalence of Marfan syndrome within the United States has been estimated to be approximately 40,000, with a 3 to 5 per 10,000 people occurrence (R. E. Pyeritz, personal communication, May 5, 1996). Males and females are equally likely to be afflicted with Marfan syndrome as are all ethnic groups. Many different mutations causing Marfan syndrome have been discovered in the fibrillin-1 gene with families sharing the same mutation. Since the disorder is autosomal dominant, individuals with Marfan syndrome have a 50% risk of transmitting the disorder to their children. However, Marfan syndrome has been found to result from spontaneous mutation (approximately 26% diagnosed). The family physician conducts an initial screening for the disorder and usually refers the patient to a medical geneticist or occasionally the cardiologist or ophthalmologist where the diagnosis is confirmed.

Major abnormalities may occur in the musculoskeletal, ocular (i.e., pertaining to the eye), and cardiovascular systems of the body. The clinical presentation of symptoms may vary

immensely, however, due to the large number of fibrillin-gene mutations documented among this population. The most serious symptoms are often associated with the cardiovascular system (95% occurrence among adults). For example, dilatation (i.e., widening) of the aorta occurs in approximately 85% of individuals with Marfan syndrome (Pyeritz & McKusick, 1979). As a result, many of these individuals are high risk for dissection (i.e., tearing) and/or sudden rupture of the aorta as they become older. As a result, surgery may be required. Another commonly occurring cardiac condition (approximately 60%) is the prolapse of the mitral valve where, in the most serious cases, blood leaks backwards through the heart valve. Symptoms stemming from prolapse of the mitral valve may include arrhythmia of the heart (i.e., irregular pulse), shortness of breath, and/or chronic fatigue. Cardiovascular complications are primarily responsible for the diminished life expectancy affiliated with the disorder (Habbal, 1992) and contribute to approximately 90% of the mortalities among individuals with Marfan syndrome (Patton, Galliani, Johnson, Hedlund, 1995). Survival well into middle age and beyond is now expected due to early diagnosis and to recent developments in medical and surgical procedures. Also the use of beta-adrenergic blocking drugs (i.e., medication that slows heart rate and lowers blood pressure), such as propranolol hydrochloride and atenolol, has recently reduced cardiovascular complications (Shores, Berger, Murphy, & Pyeritz, 1994).

Individuals with Marfan syndrome are commonly described as tall and slender. Fingers and toes (i.e., arachnodactyly) as well as arms and legs (i.e., dolichostenomelia) are often longer than normal (Manusov & Martucci, 1994). Joint laxity (i.e., loose joints) and flexion contractures (i.e., drawing together of muscles around the joints that results in deformities) are also highly prevalent. Scoliosis (i.e., abnormal curvature of the spine) and pectus deformities (e.g.,

protrusion or indentation of the sternum) are common. Overcrowded teeth caused by narrow jaws and mouths may require dental surgery. Seen more often in adults than children, striae atrophicae (i.e., stretch marks) has also been documented (Grahame & Pyeritz, 1995), but pose little, if any, health risk.

The most common ocular manifestations include ectopia lentis (i.e., dislocation of the eye lense), myopia (i.e., near-sightedness), and amblyopia (i.e., lazy eye). Ectopia lentis occurs in approximately 40 to 60% of individuals with Marfan syndrome (Maumenee, 1981; Pyeritz & Mckusick, 1979). Tearing of the retina occurs in approximately 7 to 10% of the Marfan population (Maumenee, 1981). After the age of 40, cataracts and/or acute glaucoma is common and surgical repair of dislocated lenses may be warranted. Acute or chronic glaucoma may present prior to adulthood, however.

Age-related Outcomes

Little research has been conducted exploring the educational and psychological features or co-presence of other disorders (e.g., learning disabilities, attention deficit hyperactivity disorder, depression) which may be associated with Marfan syndrome. To date, only one study (Hofman, Bernhardt, & Pyeritz, 1988) has been conducted exploring the incidence of learning disabilities and attention deficit hyperactivity disorder among children with Marfan syndrome. Based on a sample of 30 school-age children diagnosed with Marfan's, the researchers identified 13% as having a learning disability and 17% as having an attention deficit disorder. Although these results may suggest a higher incidence of learning disabilities and attention deficit disorders among individuals with Marfan syndrome compared to the general population, the authors recommended caution in generalizing the results to the entire Marfan population given the small

size of their sample and the uncertainty of the incidence of learning disabilities among the general population. In fact, no research has adequately determined whether there is a higher incidence of learning disabilities and attention deficit disorders among individuals with Marfan syndrome compared to the general population (J. G. Davis, personal communication, April 18, 1996) pointing to the need for additional research in this area.

The Hofman et al. (1988) study reported an average Wechsler Full Scale IQ of 109.3 (SD=15.4), Verbal IQ of 112 (SD=16.7) and Performance IQ of 103.8 (SD=13.8) for the 30 children with Marfan syndrome. Therefore, the vast majority of the children within the sample tended to display ability levels within the average to high average range. Nine children displayed verbal-performance discrepancies of 20 points or more. Here again, generalization to the entire Marfan population should be done cautiously. Consideration of the Verbal IQ>Performance IQ pattern is warranted, however. The authors suggested that the lower Performance IQ scores may stem from hypermobility of the wrists and/or hands. In addition, the influence of beta-adrenergic blocking drugs on the Wechsler Performance Scale subtests was suggested; however, additional research is needed to substantiate this finding.

In another study exploring the psychosocial functioning of adolescents and young adults (n=22) with Marfan syndrome, Schneider, Davis, Boxer, Fischer, and Friedman (1990) found that the average age of initial diagnosis was 10.3 years (medium age 11.5; range 6 months to 17 years). These results suggest that age of diagnosis varies and management plans, though typically focused on the prevention of life-threatening complications, need to consider the age of the individual. For example, younger children may have more difficulty restraining from physical play with peers whereas adolescents may restrain from physical exertions but feel an emotional loss for

activities they once enjoyed. Furthermore, although Schneider et al. (1990) concluded that adolescents and young adults displayed normal psychosocial adaptation, it was clear that these individuals believed their lives would be better without Marfan syndrome, they would be more physically attractive, and they would be more physically active.

Although definitive conclusions regarding the impact of Marfan syndrome upon the developmental process are lacking, specific manifestations of the syndrome at varying age levels can be logically extracted from the literature. Infants with Marfan syndrome may experience developmental delays in walking because of joint laxity (Hofman, Bernhardt, & Pyeritz, 1988). Also, dislocation of the joints, particularly the kneecaps as they begin to crawl, are common (Bernhardt, 1992). Delay in fine motor skills, due to abnormal joint flexibility of the wrists and fingers, may also interfere with learning to draw and write. Near-sightedness and amblyopia (i.e., lazy eye) may interfere with reading the blackboard. Although common among many children, poor eyesight hinders a child's ability to learn and may be mistaken for a learning problem if not found and corrected.

Children and adolescents are often taller than their same-aged peers. Therefore, it is not uncommon for them to be teased and/or expected to excel in certain sports (i.e., basketball, volleyball). Also, their height may encourage adults to expect youngsters with Marfan syndrome to be more psychologically mature than their chronological age. Finally, children and adolescents may experience growing pains from fast growth in their legs and arms, as well as other pain associated with musculoskeletal deformities (Grahame & Pyeritz, 1995).

As children with Marfan syndrome approach adolescence, they may become more self-conscious about their physical appearance and withdraw from social interactions with peers.

Depression may ensue. Rate of medical noncompliance (e.g., being more physically active than recommended, not taking medication) also tends to be relatively high during adolescence (Schneider et al., 1990). During mid to late adolescence, concerns regarding marriage, having children, and future employment options may begin to emerge. Females with Marfan syndrome may be unaware of potential problems should they become pregnant (Schneider et al., 1990). In addition, they may be uninformed regarding genetic transmission and symptom variability. Finally, future career choices will need to focus on occupations that do not require physical labor.

Psychoeducational Implications

Teachers, school psychologists, school counselors, and other professionals, within the school may assist children and adolescents with Marfan syndrome by providing academic and/or psychological support. Many children and adolescents with Marfan syndrome will require accommodations within the classroom to facilitate learning. For example, teachers may need to provide close visual presentation of materials because of near-sightedness. Hypermobility of the wrist and/or fingers may require accommodations for individuals with poor handwriting. Physical education teachers will need to develop specific exercises based upon the child's limited abilities and severity of symptoms (Romeo, 1992).

Medical attention will likely result in frequent school absences. It is common for some children to simultaneously receive treatment from a cardiologist, dentist, ophthalmologist, speech therapist, and physical therapist. The teacher will need to work closely with the child and the child's parents to ensure that he or she does not fall behind academically. For extended absences, the teacher may need to develop lessons in advance to be completed by the student as he or she

recuperates in the hospital and/or the home. In some cases, homebound instruction should be considered.

The teacher of a student with Marfan syndrome should be prepared to address possible social and emotional problems within the classroom that stem from interactions with peers. As with other disabilities, teasing by other children is common. Both the apparent physical abnormalities (e.g., long arm span, elongated lower extremities) and the covert limitations such as lack of physical stamina, put the child with Marfan at risk for peer scrutiny. These children are likely to fear undressing in public due to their distorted body-types and feel inferior (or incapable) when engaging in certain sports. In contrast, some children and adolescents may feel pressured to play and/or be expected to excel in certain sports given their height. Teachers and other school professionals will be expected to handle peer teasing and misconceptions as well as educate other students if a child with Marfan syndrome attends their school.

The school psychologist and school counselor are in unique positions to facilitate adaptation and psychological well-being. For example, counseling may be needed for depression due to physical and social complications, death of a parent who was afflicted with the syndrome, or adjustment issues that may arise once the initial diagnosis has been confirmed. The school psychologist and school counselor may also help the child and family prepare for extended hospital stays. Collaboration with medical professionals (e.g., hospital social worker, cardiac rehabilitation specialist) to facilitate transition back to school is recommended. The school psychologist should become knowledgeable about Marfan syndrome, be a resource to other professionals, and, most importantly, be available to the family and child when adjustment issues are present.

Acknowledgments

The authors gratefully acknowledge the assistance of Jessica G. Davis, MD at The New York Hospital-Cornell, Priscilla Ciccariello, Chairperson of the National Marfan Association, Reed E. Pyeritz, MD at the Institute for Medical Genetics, Medical College of Pennsylvania & Hahnemann University and the two anonymous reviewers.

Annotated Bibliography

National Marfan Foundation
382 Main Street
Port Washington, New York 11050-3121
Tel/Fax (516) 883-8712
1-(800) 8-MARFAN
http://www.marfan.org/

Bernhardt, B. (1992). The Marfan syndrome: A booklet for teachers. Port Washington, NY: National Marfan Foundation.

This booklet provides an overview of Marfan syndrome and addresses some of the most common questions asked by teachers. A brief description of the medical problems associated with Marfan syndrome and suggestions on how teachers can help children with Marfan children are provided.

Pyeritz, R. E., & Gasner, C. (1994). The Marfan Syndrome. Port Washington, NY: National Marfan Foundation.

This booklet provides an excellent overview of Marfan syndrome. Information is provided in a succinct educational format without overwhelming the reader with unnecessary medical jargon. It is especially helpful in addressing common questions related to the management and treatment of Marfan syndrome.

References

Aoyama, T. Francke, U., Gasner, C., & Furthmayr, H. (1995). Fibrillin abnormalities and prognosis in Marfan syndrome and related disorders. American Journal of Medical Genetics, 58, 169-176.

Dietz, H. C., & Pyeritz, R. E. (1995). Mutations in the human gene for fibrillin-1 (FBN1) in the Marfan syndrome and related disorders. Human Molecular Genetics, 4, 1799 -1809.

Grahame, R., & Pyeritz, R. E. (1995). The Marfan syndrome: Joint and skin manifestations are prevalent and correlated. British Journal of Rheumatology, 34, 126-131.

Habbal, M. H. E. (1992). Cardiovascular manifestations of Marfan's syndrome in the young. American Heart Journal, 123, 253-257.

Hofman, K. J., Bernhardt, B. A., & Pyeritz, R. E. (1988). Marfan syndrome: Neuropsychological aspects. American Journal of Medical Genetics, 31, 331-338.

Manusov, E. G., & Martucci, E. (1994). The Marfan syndrome: An Underground killer, Archives of Family Medicine, 3, 822-826.

Maumenee, I. H. (1981). The eye in Marfan syndrome. Trans American Ophth Soc, 79, 685-733.

Patton, D. J., Galliani, C. A., Johnson, W. H., & Hedlund, G. L. (1995). Sudden death in Marfan syndrome. American Journal of Roentgenology, 165, 160.

Pyeritz, R. E., & McKusick, V. A. (1979). The Marfan syndrome: Diagnosis and management. New England Journal of Medicine, 300, 772-777.

Romeo, T. J. (1992). The Marfan syndrome: Physical activity guidelines for physical educators, coaches, and physicians. Port Washington, NY: National Marfan Foundation.

Schneider, M. B., Davis, J. G., Boxer, R. A., Fisher, M., & Friedman, S. B. (1990). Marfan syndrome in adolescents and young adults: Psychosocial functioning and knowledge. Developmental and Behavioral Pediatrics, 11, 122-127.

Shores, J., Berger, K. R., Murphy, E. A., & Pyeritz, R. E. (1994). Progression of aortic dilatation and the benefit of long-term beta-adrenergic blockade in Marfan's syndrome. New England Journal of Medicine, 330, 1335 - 1141.

Tolliss, D. (1995). Who was...Marfan? Nursing Times, 91(23), 47.